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EPILEPSY IN A PATIENT WITH SUPERNUMERARY MOSAIC RING CHROMOSOME 19

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Patient with supernumerary small ring chromosome are rare. To date, only four cases have been described with a ring chromosome derived from 19, only one with febrile convulsions (FC).
The authors report a mentally retarded overweight boy aged 16 years 8 months, showing macrocephaly, hypertelorism, anti-mongoloid slants and epilepsy. He started to present convulsive seizures with or without hyperthermia at the age of one year. Then hemiclonic and complex partial seizures preceded by an epigastric aura with or without secondary generalization appeared.
EEG tracings showed diffuse and multifocal spike-wave discharges over the left fronto-central, temporal and the right fronto-central regions.
Cerebral MRI was normal.
The patient showed a mosaic karyotype with at least two different kinds of de novo ring chromosomes 19 identified by fluorescent in situ hybridization (FISH) with probe D1Z7/D5Z2/D19Z3 and painting specific for chromosme 19. To determine the genetic content of the rings FISH was performed with cosmids specific for p and q arms of chromosome 19; the small rings extend from the centromere to 19p12 while the large rings from the centromere to 19q12. A more detailed analysis of the genetic content is in progress.

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